INSTITUTE OF IMMUNOLOGY AND GENETICS

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Requesting physician:	**Patient information:**

Request Form Connective tissue and vascular diseases

Cause of examination: ☐ diagnostic ☐ predictive for familial mutation _____

Clinical information:

Family history:

(Check all that apply)

☐ Patient clinically affected

☐ No previous molecular genetic examinations existent

☐ The following previous molecular genetic examinations have been done:

_____

Date / Time of sampling:	**Signature:**

Specimen requirements and logistics:

- **Patient Consent Form for Genetic Testing according to the German Law and Request Form**
- **Billing information (Insurance-, Institutional- or Selfpay-Billing)**
- **5 ml EDTA blood collection tube labeled with patient name and date of birth**

Patient name: _____ **DOB:** _____

☐ **Loeys-Dietz-syndrome**
Stufe 1: *SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2*
Stufe 2: *TGFBR2 (MLPA)*

☐ **Marfan-syndrome**
Stufe 1: *FBN1*
Stufe 2: FBN1 (MLPA)

☐ **Shprintzen-Goldberg-syndrome**
SKI

☐ **Familial thoracic aortic aneurysm and dissection**
ACTA2, BGN, COL3A1, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, PRKG1, FBN1, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2

☐ **Ehlers-Danlos-syndrome, autosomal-dominant**
C1R, C1S, COL1A1, COL5A1, COL5A2, COL1A2, COL3A1

☐ **Ehlers-Danlos-syndrome, autosomal-recessive**
ADAMTS2, B3GALT6, B4GALT7, SLC39A13, *CHST14, DSE, FKBP14, PLOD1*

☐ **Stickler-syndrome**
COL2A1, COL11A1, COL11A2, COL9A1, COL9A2, COL9A3

☐ **Cutis laxa**
ELN, *FBLN5*, *ATP6V0A2*, *ALDH18A1*, *EFEMP2*, *PYCR1*, *LTBP4*, *ATP6V1A, RIN2*

☐ **Osteogenesis imperfecta**
Stufe 1: *COL1A1, COL1A2*
Stufe 2: *BMP1, CRTAP, FKBP10, P3H1, PLOD2, PPIB, SEC24D, SERPINH1, SPARC, TMEM38B, CREB3L1, IFITM5, MBTPS2, MESD, SERPINF1, SP7, TENT5A, WNT1*

☐ **Various connective tissue disorders**
ABCC6, COL2A1, FBN2, GORAB, PLOD3